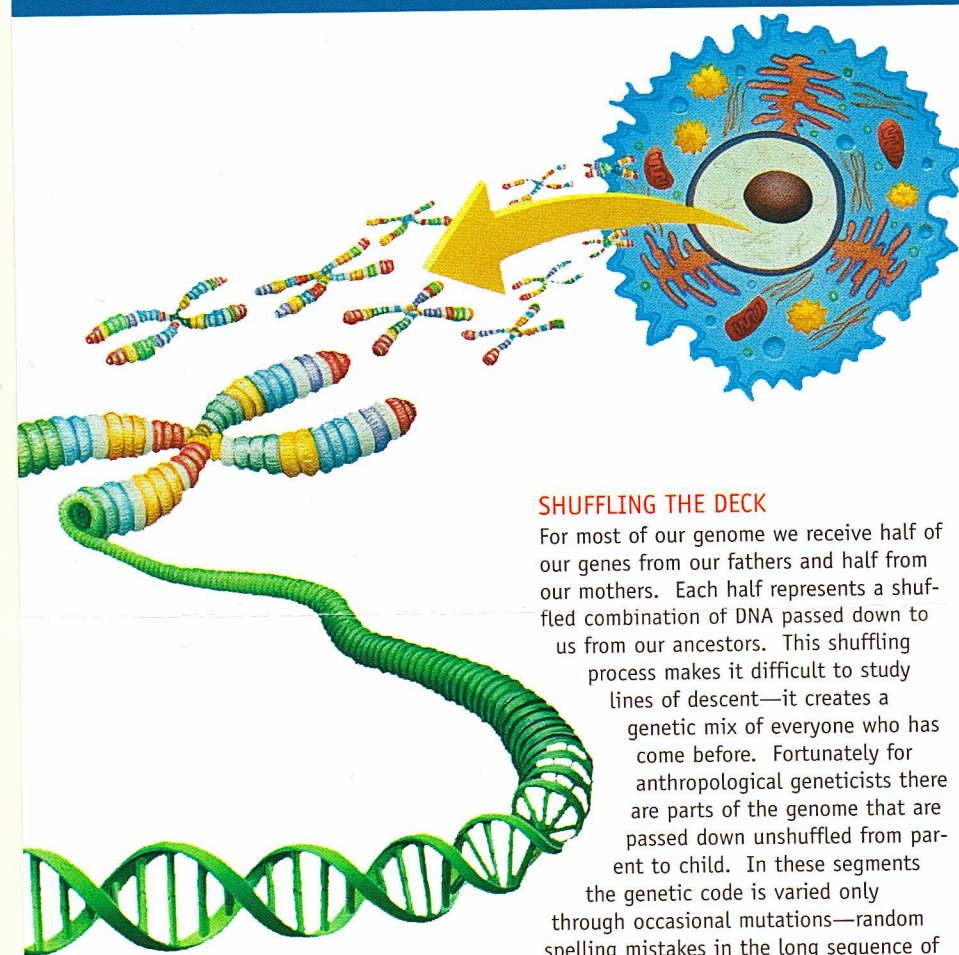


The How and Why of DNA: A Genetics Primer

What is Anthropological Genetics?



SHUFFLING THE DECK

For most of our genome we receive half of our genes from our fathers and half from our mothers. Each half represents a shuffled combination of DNA passed down to us from our ancestors. This shuffling process makes it difficult to study lines of descent—it creates a genetic mix of everyone who has come before. Fortunately for anthropological geneticists there are parts of the genome that are passed down unshuffled from parent to child. In these segments the genetic code is varied only through occasional mutations—random spelling mistakes in the long sequence of letters that make up our DNA. When these mutations are passed down through the generations they become markers of descent.

GENETIC SIGNPOSTS

By following a marker in these unshuffled regions back through the generations to its origin, geneticists can pinpoint the most recent common ancestor of everyone alive who carries that marker. In this way markers serve as evolutionary signposts that define different human lineages.

These signposts allow the disparate branches of the human family tree to be followed back through time to an African root—a common human ancestor who gave rise to all of the genetic lineages we see in the world today.

THE Y CHROMOSOME

Y-chromosomal DNA, passed from father to son, is one of these highly effective genetic tools for tracing human lineages through history. But this clarity comes at a price—tracing its genetic path yields no record of the female half of the evolutionary equation. The absence of female representation leaves our genetic heritage incomplete, in need of another segment of DNA that can fill this void.

MITOCHONDRIAL DNA

Mitochondrial DNA is passed from mother to offspring, allowing us to trace a female line of descent. Although it is much smaller than the Y chromosome, the mitochondrial genome mutates at a higher rate than the rest of our DNA. Over time this has created an abundant amount of variation around the world to study.

Both DNA types have advantages and can be used together to form a more complete picture of our genealogical past. That picture has now begun to take an incredible shape.

A COMMON ROOT

Geneticist Spencer Wells and his colleagues have defined part of the human journey using Y-chromosome data and determined that the common ancestor of every man alive was an African, dubbed "Adam", who lived around 60,000 years ago. Other researchers have found similar evidence of a common African origin for mitochondrial DNA—"Eve" to our Y-chromosome Adam.

Adam and Eve were not the only humans alive at the time, only the luckiest. Their genetic lineages are the only ones that did not die out over the ages. By following the distinctive genetic signatures of their descendants we can trace the human journey out of Africa and around the world, and see that it may have happened much more quickly, and recently, than many supposed.

THE JOURNEY

Genetic data indicate that the first great migration out of Africa was along Asia's southern coastline, through India and Southeast Asia and on to Australia—linking that country's aboriginal people back to Africa within the last 50,000 years.

Forty-five thousand years ago more of Adam and Eve's descendants reached the Middle East, and as the ice age waned some 40,000 years ago they moved into the open steppes of Central Asia.

If Africa is the cradle of humanity, Central Asia served as its nursery. From these plentiful steppes early humans multiplied and set out to populate much of the globe. Around 35,000 years ago they reached Europe. Some 20,000 years ago a separate group of Central Asians moved north into Siberia and the Arctic Circle. From there, perhaps 5,000 years later, they crossed into the Americas.

Many archaeologists place the first wave of "Out of Africa" migration at around 100,000 years ago. This time discrepancy awaits explanation, as does the original impetus for migration. It may have been spurred by booming populations and food competition, or by climate change. Some scientists believe that a genetic mutation during this era increased human cognitive powers and may have led to the development of language.

Whatever their reasons for leaving Africa, humans adapted over time to their new environments. They grew paler, for example, in areas with less sunlight and acquired many

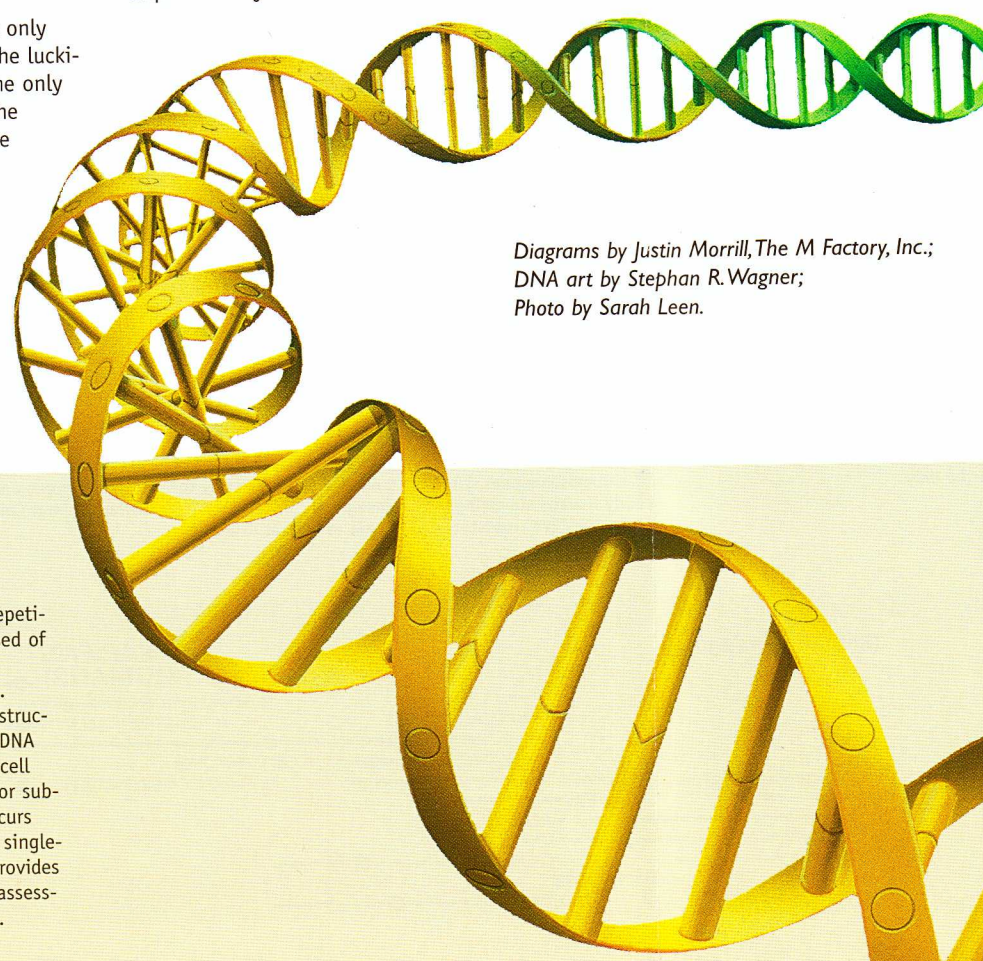
physical characteristics that now distinguish different peoples from around the globe.

The tale is a remarkable one. The descendants of a small group of Africans, multiplying over time, split into groups defined by their own genetic markers and spread across the planet.

UNRAVELING THE MYSTERY

But the story is far from complete. As Wells says, "We may have a view of the forest but we still know very little about the trees." Geneticists hope to track markers more comprehensively throughout all human populations and create a far more detailed map of the human journey. To do so they must greatly expand the pool of samples available from around the world, particularly from indigenous peoples, who retain a clearer picture of the geographic context in which the genetic lineages arose. Reaching such people, and convincing them to share their genetic stories, is urgent in a world where local cultural identities are now endangered.

Our genes have a remarkable tale to tell, but they can't do it alone. Paleoclimatology, linguistics, archaeology, and other sciences have critical roles to play in interpreting the human journey from our common ancestors to the present day. But the outline of the story has been with us all along. The DNA molecule, sometimes called the "secret of life," also holds the secrets of humanity's incredible journey.



Diagrams by Justin Morrill, The M Factory, Inc.;
DNA art by Stephan R. Wagner;
Photo by Sarah Leen.

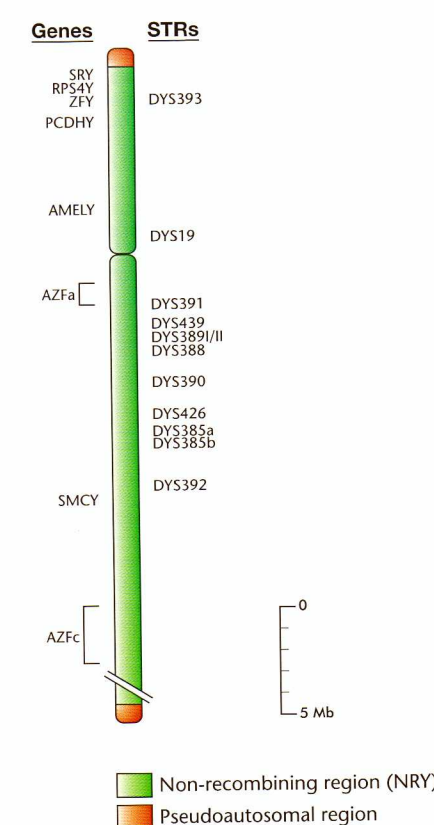
The Genetic Signposts

MALE LINEAGE

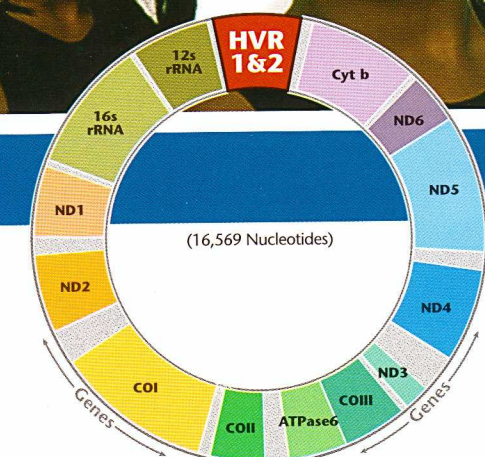
The Y Chromosome

The Y chromosome is the sex-determining chromosome in humans. While all other chromosomes are found in matching pairs, it is the mismatch of the Y with its partner, the X chromosome, that determines gender—men have a mismatched pair (Y and X), while women have two X chromosomes. Because the Y does not have a matching chromosome, most of it (the non-recombining region, or NRY) escapes the shuffling process known as recombination that occurs every generation in the rest of our genome. This allows the Y to be passed down through a purely male line, changed only by random mutational events. These mutations have given rise to many useful genetic markers, such as those shown on the tree diagram to the right (e.g., M91, M60, etc.), with varying geographic distributions in men from around the world.

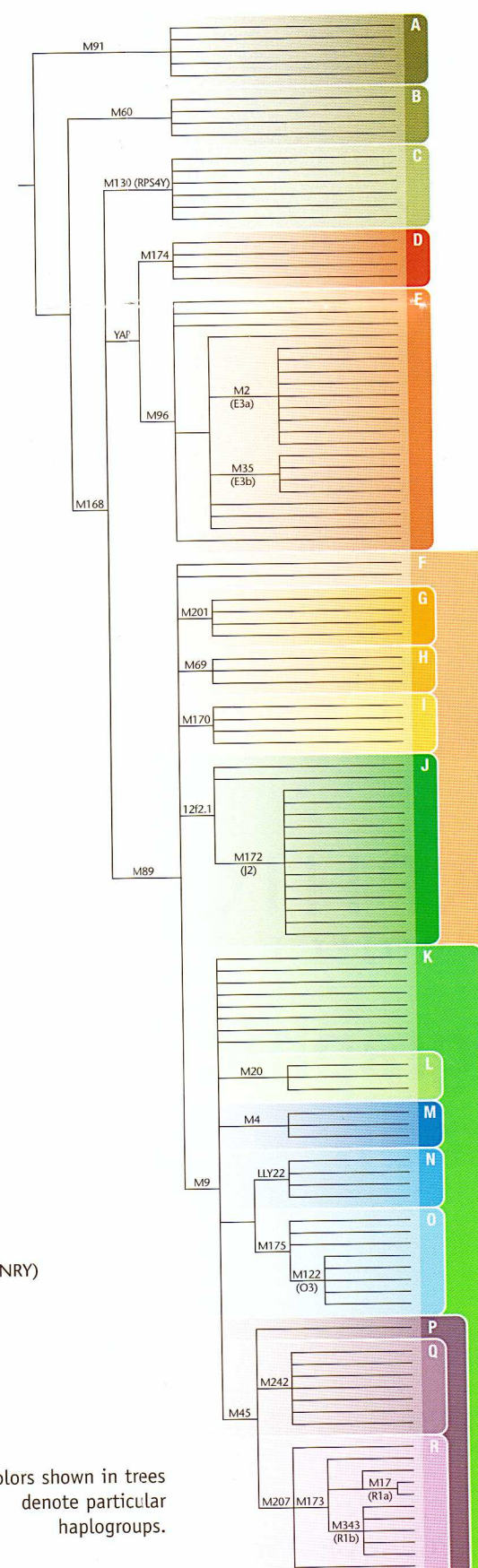
Y CHROMOSOME



mtDNA GENOME



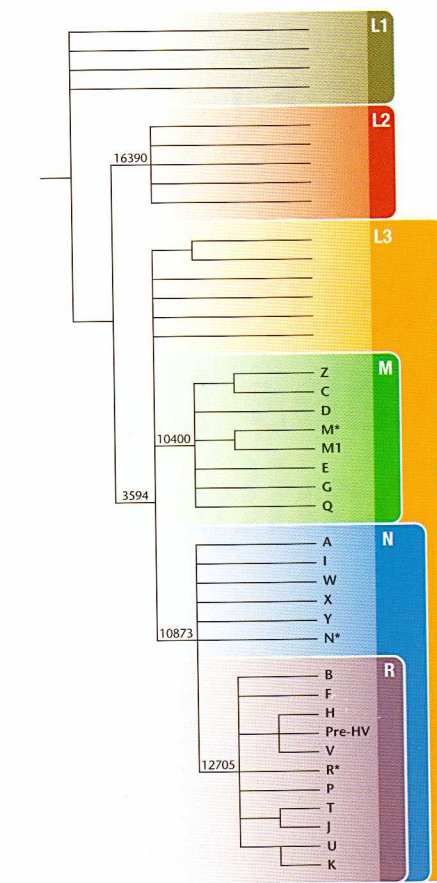
Y CHROMOSOME TREE



FEMALE LINEAGE Mitochondrial DNA

If the Y chromosome traces the male lineage back through history, then the mitochondrial genome (mtDNA) can be considered its female counterpart. Mitochondria are self-reproducing structures found inside the cells of all higher organisms, typically present in hundreds of copies per cell. They are responsible for generating most of the energy used by the cell. Because there are no mitochondria in the head of a mature sperm, they are passed down solely from mother to offspring. One region of particular importance in mtDNA is the hypervariable region (HVR 1&2), where the rate of mutation has been shown to be up to 100 times greater than that of the nuclear genome. Because of its much shorter length (several hundred nucleotides, versus millions of nucleotides, or Mb, for the Y), the HVR can be quickly scanned to reveal many informative mutational events that have been passed down through the maternal line.

mtDNA TREE



Glossary

DNA

Containing all genetic information, DNA is a long, linear sequence of small molecules called nucleotides. There are four different nucleotides that constitute the double-helical DNA strand, denoted by the symbols A, C, T, and G. These are often referred to as the "building blocks of life," and their specific order and position in the genome encodes the information carried by the DNA. DNA stands for DeoxyriboNucleic Acid.

Mutation

A random change in the DNA sequence that occurs as a copying mistake during cell division. It may be the replacement of a single nucleotide with another, or the addition or deletion of nucleotides. Mutations are very rare, but given the large size of the human genome (approx. six billion nucleotides) children have around fifty single-nucleotide mutations that distinguish them from their parents.

Haplogroup (Hg)

A group of genetic lineages that share a mutation or set of mutations specific to individuals in that group. Because haplogroups are defined by mutational events that occurred at a specific place and time in history, they typically have geographic distributions that are representative of the age and scope of migration events that make up that history.

STR

Short Tandem Repeats, also called microsatellites, are repetitive DNA sequences composed of units a few nucleotides in length—e.g., CTCTCTCT. . . . Because of their repetitive structure, STRs can confuse the DNA copying mechanism during cell division, causing it to add or subtract a repeat unit. This occurs much more frequently than single-nucleotide mutation, and provides geneticists with a tool for assessing the age of haplogroups.

Colors shown in trees denote particular haplogroups.

Populating the World: Tracing the Journey of our Ancestors

HUMANS VERSUS NEANDERTALS?
Are Neandertals the ancestors of modern Europeans? Geneticists say no. Mitochondrial DNA recently obtained from Neandertal bones reveals a very different genetic sequence. The true European ancestors were the tall, lithe newcomers, bearing the genetic marker M173 on their Y chromosomes, who arrived after generations spent on the Central Asian steppes. Did they kill off the stocky, muscular Neandertals they encountered? Not likely. As more efficient hunters with better tools and highly cooperative societies, they probably outcompeted their predecessors, gradually pre-empting the food supply. Within a few thousand years the Neandertals had dwindled and vanished, leaving no living trace behind.

1 African Origins
Fossils indicate that modern *Homo sapiens* evolved 200,000 years ago in the highlands of East Africa. Geneticists agree, tracing the mitochondrial DNA of everyone alive today back to an ancestral female who lived in the region roughly 150,000 years ago. This "Eve" is mirrored by an "Adam" who dwelt on those same savannas about 60,000 years ago, bequeathing to every man alive today a common Y chromosome legacy. Of the several hominid species then existing, including other bands of early humans, people today probably spring from just one small tribe of hunter-gatherers, perhaps a little smarter, more linguistically advanced, and possessing better tools than their neighbors.

2 Out of Africa: Y Chromosome Evidence
We were not the first hominids to leave Africa. Earlier species had already spread over Eurasia when our distant ancestors began their own journey nearly 60,000 years ago. Genetic markers on the Y chromosome offer the clearest evidence of that great migration's various stages. (These markers are denoted by M followed by numbers that do not correspond to chronological sequence.) Early mutations (M91, M60) on the Y chromosome rootstock bequeathed by "Adam" are found in modern African males, reflecting migrations from East Africa as his descendants fanned out across that continent's savannas and forests. Soon thereafter, another mutation (M168) arose in a man probably living in northeast Africa—and the lineage he fathered colonized the rest of the world. One such (called "YAP") gave rise to further markers (M96, M174) apparently reflecting subsequent movements in Africa, the Mediterranean, and East Asia, but its incidence is rare today in most non-African populations.

Not so another early mutation, known as M130. Found principally in Southeast Asian and Australian Aboriginal males, it reflects what may be the earliest substantial migration out of Africa, occurring more than 50,000 years ago. Sea levels were lower then, and these beachcombing humans probably followed the long sandy coasts of Arabia, India, and Southeast Asia—leaving genetic traces in populations there today—until they eventually reached Australia.

MIGRATION PATTERNS OF EARLY HUMANS		
Y Chromosome Markers... (thousand years ago)		
M 91.....60	M 9.....40	M 201.....20
M 60.....50	M 175.....35	M 242.....20
M 168.....50	M 45.....35	M 3.....10
YAP.....50	M 173.....30	M 172.....10
M 174.....50	M 20.....30	M 17.....10
M 130.....50	M 69.....30	M 122.....10
M 96.....45	M 170.....25	M 4.....10
M 89.....45	M 2.....20	LLY 22.....10
mtDNA Markers... (thousand years ago)		
L 1.....>100	F.....50	Z.....30
L 2.....80	R.....50	HV.....30
L 3.....70	D.....45	H.....30
M.....60	pre-HV.....40	N1.....30
M1.....60	U.....40	I.....30
N.....50	K.....40	X.....30
B.....50	JT.....40	W.....30
C.....50	A.....30	V.....15

Neandertal skull and Modern Human skull photos by Kenneth Garrett; Mungo Skull photo by Wilfred Shawcross; Omo Skull photo "Courtesy of the Natural History Museum London"; Kennewick Skull photo by Chip Clark; Cast by James C. Chatters; Hobbit Skull art by Greg Hartlin; Relief by Tibor Tóth.
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THE GENOGRAPHIC PROJECT

National Geographic Maps the Human Family Tree

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The Hominid Family Tree

Although the hominid family tree cannot be described in detail, general outlines are apparent. Some extremely ancient fossils, such as *Orrorin*, six million years old, resemble the great apes but appear to have been bipedal, the signature trait of hominids. Nearly four million years ago the Australopithecines

first appeared. They may—or may not—have given rise, over two million years later, to the genus *Homo*, the human family. Equipped with larger brains and better hand tools, early humans were the first hominids to leave Africa and explore the world. At least four distinct species, ranging from the burly

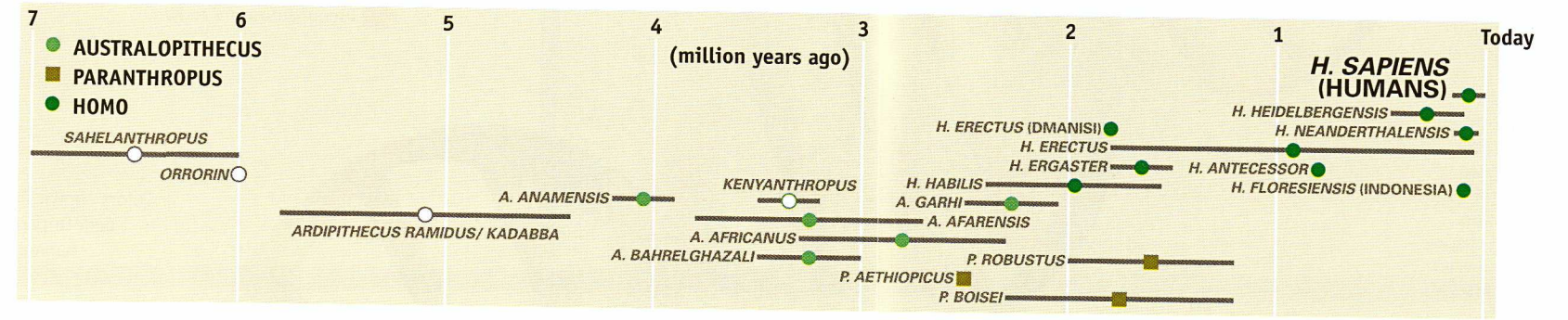
Homo neanderthalensis of Europe to the elfin *Homo floresiensis* of Southeast Asia, still survived at the time our ancestors, *Homo sapiens*, emerged on the scene. We proved more adaptable and resilient, and today are the only hominids left.

The Mitochondrial DNA Evidence

Footprints left by mitochondrial DNA (mtDNA) are not as clearly defined as those left by the Y chromosome. Nevertheless, mtDNA lineages, each tagged with a letter of the alphabet, broadly shadow Y chromosome ones. L3, an early lineage stemming from the "Eve" rootstock,

probably appeared in northeast Africa around 70,000 years ago. There it branched, reflecting two great dispersals of humanity. One is traced by the lineage known as M, which points toward Australia but gave rise along the way to lineages (C, D, Z), also found in Central Asia and

Siberia. The other is traced by the lineage N, reflecting movement up through the Middle East into Eurasia, where subsequent lineages (X, A, B, and R among them) branch north toward the Arctic, west toward Europe, and east toward central China.



A Genetic Odyssey

How DID HUMANS COME TO POPULATE THE WORLD? For years paleoanthropologists and archaeologists sought the answer in the only clues they could find, the scattered remains of our distant ancestors. Yet ancient bones and artifacts could only tell a fragmentary tale. Today, however, geneticists have discovered complete records of prehistoric migration inscribed deep within our genes. Mutations in certain components of DNA have accumulated in sequences like the layers of an archaeological dig. By comparing these sequences across global populations, and then plotting the results on a map, scientists have revealed the broad outlines of early human movements—the genetic footprints left by our ancestors as they ventured out of Africa to colonize the world. Long, interlacing arrows have been charted, each marking the passage of a distinct genetic lineage. Hugging coastlines, skirting deserts, drifting across grasslands, these people wandered in a seasonal ebb and flow, generation after generation, following the rhythms of hunting and gathering. Progress was imperceptible, but after 50,000 years humans had occupied every habitable continent on Earth. Along the way they gave rise to the diverse populations found around the world today.